

WHAT IS CLAIMED IS:

1. An isolated stem cell or stem cell line carrying a disease-causing mutation in a genomic polynucleotide sequence thereof.
2. The isolated stem cell or stem cell line of claim 1, wherein said stem cell is of embryonic origin.
3. The isolated stem cell or stem cell line of claim 1, wherein said stem cell is of human origin.
4. The isolated stem cell or stem cell line of claim 1, wherein said disease-causing mutation is selected from the group consisting of a missense mutation, a nonsense mutation, a frameshift mutation, a readthrough mutation, a promoter mutation, a regulatory mutation, a deletion, an insertion, an inversion, a splice mutation and a duplication.
5. The isolated stem cell or stem cell line of claim 1, wherein said disease-causing mutation is associated with a genetic disorder selected from the group consisting of cystic fibrosis (CF), myotonic dystrophy (DM), van Waardenburg syndrome (WS), metachromatic leukodystrophy (MLD), Gorlin disease, Huntington's disease (HD), spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD).
6. The isolated stem cell or stem cell line of claim 1, wherein said disease-causing mutation is selected from the group consisting of the W1282X as set forth in SEQ ID NO:24 associated with cystic fibrosis, the PAX3-del28 (510del28 in SEQ ID NO:34) associated with van Waardenburg syndrome, more than 50 (CTG) repeats as set forth in SEQ ID NO:22 associated with Myotonic dystrophy and the 1505C→T (P377L) as set forth in SEQ ID NO:21 associated with metachromatic leukodystrophy.

7. The isolated stem cell or stem cell line of claim 1, wherein said stem cell is capable of being maintained in an undifferentiated state for at least 41 passages.

8. The isolated stem cell or stem cell line of claim 1, wherein said stem cell exhibits a karyotype of 46, XX or 46, XY following at least 30 passages.

9. The isolated stem cell or stem cell line of claim 1, wherein said stem cell exhibits pluripotent capacity following 40 passages.

10. The isolated stem cell or stem cell line of claim 1, wherein the stem cell is suspended in a culture medium including serum or serum replacement.

11. The isolated stem cell of claim 10, wherein said serum is provided at a concentration of at least 10 % and said serum replacement is provided at a concentration of at least 15 %.

12. An isolated embryoid body comprising a plurality of cells at least some of which carry a disease-causing mutation in a genomic polynucleotide sequence thereof.

13. The isolated embryoid body of claim 12, wherein said embryoid body is derived from a stem cell or a stem cell line.

14. The isolated embryoid body of claim 13, wherein said stem cell is of embryonic origin.

15. The isolated embryoid body of claim 13, wherein said stem cell is of human origin.

16. The isolated stem cell or stem cell line of claim 13, wherein said stem cell exhibits a karyotype of 46, XX or 46, XY following at least 30 passages.

17. The isolated embryoid body of claim 12, wherein said disease-causing mutation is selected from the group consisting of a missense mutation, a nonsense mutation, a frameshift mutation, a readthrough mutation, a promoter mutation, a regulatory mutation, a deletion, an insertion, an inversion, a splice mutation and a duplication.

18. The isolated embryoid body of claim 12, wherein said disease-causing mutation is associated with a genetic disorder selected from the group consisting of cystic fibrosis (CF), myotonic dystrophy (DM), van Waardenburg syndrome (WS), metachromatic leukodystrophy (MLD), Gorlin disease, Huntington's disease (HD), spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD).

19. The isolated embryoid body of claim 12, wherein said disease-causing mutation is selected from the group consisting of the W1282X as set forth in SEQ ID NO:24 associated with cystic fibrosis, the PAX3-del28 (510del28 in SEQ ID NO:34) associated with van Waardenburg syndrome, more than 50 (CTG) repeats as set forth in SEQ ID NO:22 associated with Myotonic dystrophy and the 1505C→T (P377L) as set forth in SEQ ID NO:21 associated with metachromatic leukodystrophy.

20. The isolated embryoid body of claim 12, wherein said embryoid body is capable of differentiating into cells of the embryonic ectoderm, embryonic endoderm and/or embryonic mesoderm.

21. The isolated embryoid body of claim 20, wherein said cells of the embryonic ectoderm are selected from the group consisting of neural cells, retina cells and epidermal cells.

22. The isolated embryoid body of claim 20, wherein said cells of the embryonic endoderm are selected from the group consisting of hepatocytes and pancreatic cells and secreting cells.

23. The isolated embryoid body of claim 20, wherein said cells of the embryonic mesoderm are selected from the group consisting of osseous cells, cartilaginous cells, elastic cells, fibrous cells, myocytes, myocardial cells, bone marrow cells, endothelial cells, smooth muscle cells, and hematopoietic cells.

24. The isolated embryoid body of claim 12, wherein the embryoid body is suspended in a culture medium including serum or serum replacement.

25. The isolated embryoid body of claim 24, wherein said serum is provided at a concentration of at least 10 % and said serum replacement is provided at a concentration of at least 15 %.

26. The isolated embryoid body of claim 12, wherein said embryoid body is at least 1 day-old.

27. An isolated differentiated cell, tissue or organ carrying at least one disease-causing mutation in a genomic polynucleotide sequence thereof.

28. The isolated differentiated cell, tissue or organ of claim 27, wherein said differentiated cell is selected from the group consisting of neural cells, retina cells, epidermal cells, hepatocytes, pancreatic cells, osseous cells, cartilaginous cells, elastic cells, fibrous cells, myocytes, myocardial cells, bone marrow cells, endothelial cells, smooth muscle cells, and hematopoietic cells.

29. The isolated differentiated cell, tissue or organ of claim 27, wherein said tissue is selected from the group consisting of brain tissue, retina, skin tissue, hepatic tissue, pancreatic tissue, bone, cartilage, connective tissue, muscle tissue, cardiac tissue brain tissue, vascular tissue, fat tissue, hematopoietic tissue, renal tissue, pulmonary tissue, and gonadal tissue.

30. The isolated differentiated cell, tissue or organ of claim 27, wherein said organ is selected from the group consisting of head, eye, leg, hand, heart, liver kidney, lung, pancreas, ovary, testis, brain and stomach.

31. The isolated differentiated cell, tissue or organ of claim 27, wherein said differentiated cell, tissue or organ is of human origin.

32. The isolated differentiated cell, tissue or organ of claim 27, wherein said disease-causing mutation is selected from the group consisting of a missense mutation, a nonsense mutation, a frameshift mutation, a readthrough mutation, a promoter mutation, a regulatory mutation, a deletion, an insertion, an inversion, a splice mutation and a duplication.

33. The isolated differentiated cell, tissue or organ of claim 27, wherein said disease-causing mutation is associated with a genetic disorder selected from the group consisting of cystic fibrosis (CF), myotonic dystrophy (DM), van Waardenburg syndrome (WS), metachromatic leukodystrophy (MLD), Gorlin disease, Huntington's disease (HD), spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD).

34. The isolated differentiated cell, tissue or organ of claim 27, wherein said disease-causing mutation is selected from the group consisting of the W1282X as set forth in SEQ ID NO:24 associated with cystic fibrosis, the PAX3-del28 (510del28 in SEQ ID NO:34) associated with van Waardenburg syndrome, more than 50 (CTG) repeats as set forth in SEQ ID NO:22 associated with Myotonic dystrophy and the 1505C→T (P377L) as set forth in SEQ ID NO:21 associated with metachromatic leukodystrophy.

35. A method of identifying an agent suitable for treating a disorder associated with at least one disease-causing mutation, comprising:

- (a) generating a stem cell line or an embryoid body carrying the at least one disease-causing mutation;
- (b) subjecting cells of said stem cell line or said embryoid body to differentiating conditions to thereby obtain differentiated cells exhibiting an effect of the at least one disease-causing mutation and;

(c) exposing said differentiated cells to a plurality of molecules and identifying from said plurality of molecules at least one molecule capable of regulating said effect of the at least one disease-causing mutation on said differentiated cells, said at least one molecule being the agent suitable for treating the disorder associated with the at least one disease-causing-mutation.

36. The method of claim 35, wherein said embryoid body is derived from a stem cell or a stem cell line.

37. The method of claim 35, wherein said stem cell is of embryonic origin.

38. The method of claim 35, wherein said stem cell is of human origin.

39. The method of claim 35, wherein said stem cell exhibits a karyotype of 46, XX or 46, XY following at least 30 passages.

40. The method of claim 35, wherein said disease-causing mutation is selected from the group consisting of a missense mutation, a nonsense mutation, a frameshift mutation, a readthrough mutation, a promoter mutation, a regulatory mutation, a deletion, an insertion, an inversion, a splice mutation and a duplication.

41. The method of claim 35, wherein said disease-causing mutation is associated with a genetic disorder selected from the group consisting of cystic fibrosis (CF), myotonic dystrophy (DM), van Waardenburg syndrome (WS), metachromatic leukodystrophy (MLD), Gorlin disease, Huntington's disease (HD), spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD).

42. The method of claim 35, wherein said disease-causing mutation is selected from the group consisting of the W1282X as set forth in SEQ ID NO:24 associated with cystic fibrosis, the PAX3-del28 (510del28 in SEQ ID NO:34) associated with van Waardenburg syndrome, more than 50 (CTG) repeats as set forth

in SEQ ID NO:22 associated with Myotonic dystrophy and the 1505C→T (P377L) as set forth in SEQ ID NO:21 associated with metachromatic leukodystrophy.

43. The method of claim 35, further comprising a step of isolating lineage specific cells from said embryoid body prior to step (b).

44. The method of claim 43, wherein said isolating lineage specific cells is effected by sorting of cells contained within said embryoid body via fluorescence activated cell sorter.

45. The method of claim 43, wherein said isolating lineage specific cells is effected by a mechanical separation of cells, tissues and/or tissue-like structures contained within said embryoid body.

46. The method of claim 43, wherein said lineage specific cells are of the embryonic ectoderm and are selected from the group consisting of neural cells, retina cells and epidermal cells.

47. The method of claim 43, wherein said lineage specific cells are of the embryonic endoderm and are selected from the group consisting of hepatocytes, pancreatic cells and secreting cells.

48. The method of claim 43, wherein said lineage specific cells are of the embryonic mesoderm and are selected from the group consisting of osseous cells, cartilaginous cells, elastic cells, fibrous cells, myocytes, myocardial cells, bone marrow cells, endothelial cells, smooth muscle cells, and hematopoietic cells.

49. The method of claim 35, wherein said stem cell or said embryoid body is suspended in a culture medium including serum or serum replacement.

50. The method of claim 49, wherein said serum is provided at a concentration of at least 10 % and said serum replacement is provided at a concentration of at least 15 %.

51. The method of claim 35, wherein said embryoid body is at least 1 day old.